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ARTICLE

Parents' decision-making regarding whether to receive adult-onset only genetic findings for their children: Findings from the BabySeq Project

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ABSTRACT

Purpose: Most professional guidelines recommend against genetic screening for adult-onset only (AO) conditions until adulthood, yet others argue that there may be benefit to disclosing such results. We explored parents' decision-making on this issue in the BabySeq Project, a clinical trial of newborn genomic sequencing.

Methods: We conducted interviews with parents ($N = 24$) who were given the option to receive actionable AO results for their children. Interviews explored parents' motivations to receive and reasons to decline AO genetic disease risk information, their decision-making process, and their suggestions for supporting parents in making this decision.

Results: Parents noted several motivations to receive and reasons to decline AO results. Most commonly, parents cited early intervention/surveillance ($n = 11$), implications for family health ($n = 7$), and the ability to prepare ($n = 6$) as motivations to receive these results. The most common reasons to decline were protection of the child's future autonomy ($n = 4$), negative effect on parenting ($n = 3$), and anxiety about future disease ($n = 3$). Parents identified a number of ways to support parents in making this decision.

Conclusion: Results show considerations to better support parental decision-making that aligns with their values when offering AO genetic information because it is more commonly integrated into pediatric clinical care.

Introduction

Integrating genomic sequencing into the routine clinical care of newborns may provide health benefits across the lifespan, yet it raises a number of ethical questions. One of the most debated questions around the use of genomic sequencing for minors is whether results for risk of adult-onset only (AO) conditions should be disclosed to parents. Most professional guidelines recommend against genetic screening for AO conditions until adulthood.[1-5] These guidelines appeal to the best interests of the child and argue that the potential risks, including psychosocial harm to the family and potential for

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future discrimination by insurers, outweigh the potential benefits because there are no actions to be taken in the childhood period to prevent or reduce the risk of the condition. In addition, many argue that testing for AO conditions infringes on the child's "right to an open future"[6] by limiting their future ability to make an autonomous decision about testing and results disclosure.

This debate has been reinvigorated because genomic sequencing is more commonly integrated into pediatric clinical care. For example, when genomic sequencing is provided clinically, the American College of Medical Genetics and Genomics (ACMG) recommends that clinicians disclose a specific list of clinically actionable findings to patients when found secondarily to testing indication, regardless of age. These recommendations argue that the potential for benefit to the family, such as identifying at-risk relatives, should be considered within the best interests of the minor (eg, by promoting the health of their parents). Others have suggested that disclosing such results to families can inform parents' reproductive plans and the child's future life planning, psychologically benefit the child by decreasing uncertainty and anxiety, and reduce future harm by identifying risk early.[7] The ACMG recommendations conclude that these potential benefits outweigh the risks to the child in the context of clinical genomic sequencing, but explicitly exclude the context of newborn sequencing, calling for additional data in this population.[8,9]

Studies that have assessed parents' hypothetical perspectives toward or actual decisions about receiving AO findings for their children have not found consensus among parents on whether to receive such results, with parents identifying both potential benefits and risks.[10-19] One study found that parents chose to receive AO results despite their ambivalence because of a sense of duty to learn all available information about their child's health.[10] Another found that parents generally did not think of secondary findings as different from any other health information, even if those findings were related to AO conditions.[20] No study to date has asked parents who have made this decision what suggestions they have for supporting parents making this decision. We conducted interviews with parents who were given the option to receive actionable AO results for their children as part of the BabySeq Project, a clinical trial of newborn genomic sequencing. In this article we report parents' motivations to receive and reasons to decline adult-onset genetic disease risk information for their children, their decision-making process, and their suggestions for supporting parents in making this decision.

Materials and Methods

The BabySeq Project

Study methods have been described in detail elsewhere.[21] In brief, we enrolled newborns and their parents from well-baby nurseries and intensive care units from 3 Boston-area hospitals. Babies were randomized to receive either a family history assessment and review of standard state-mandated newborn screening results or the same plus exome sequencing. Initially, exome sequencing results reported to families included dominant or biallelic recessive variants in a single gene known to significantly increase the risk of developing a condition during childhood (childhood monogenic disease risk), recessive variants, and pharmacogenomic variants associated with a limited panel of medications used in pediatrics. Results associated with more than 1000 conditions that could present and/or for which surveillance was recommended during childhood were reported.[22] If monogenic disease risk was identified in the newborn, parental DNA was genotyped to determine whether the variant was de novo or inherited and whether multiple variants for recessive traits were in cis or in trans. Inheritance was reported to the parents. Genetic results reported in the BabySeq Project have been described elsewhere (unpublished data, Green RC, Shah N, Genetti CA, et al. https://doi.org/10.1101/2022.03.18.22272284).[23,24]

Later, the study protocol was modified to allow return of results related to highly actionable AO conditions.[25] This was defined as AO conditions meeting criteria used to generate the 2013 ACMG list,[9] and in practice at the time included hereditary breast and ovarian cancer, Lynch syndrome, and MYH-related syndromes (the remaining conditions on the list could be of childhood onset and were already included in the results that were returned). Parents who enrolled in the study before AO results were routinely included in the screening were contacted and offered the opportunity to reconsent to learn if their child was at risk for actionable AO conditions identified by the study. These parents were mailed a letter about AO results with an updated consent form. Parents then participated in an informed consent session via telephone with a genetic counselor who described the risks and benefits of this expanded analysis.

Interviews

Families who made a decision about whether to receive AO results for their children were contacted with an invitation to participate in an interview about that decision. We invited all families who declined to receive adult-onset results ($n = 8$), families who both consented and received a positive adult-onset result ($n = 2$), and a random sample of families who consented and received negative adult-onset results ($n = 66$). Given the larger size of this last group, we selected among the families by using a random number generator to balance the sample. We continued to invite families who received a negative result until thematic saturation[26] was reached.

Interviews were semistructured and were conducted by a trained interviewer (S.P. or R.L.H.) over the phone or via secure video conferencing. Interviews took place an average of 18 months (range = 10-24 months) after making the decision whether to receive AO risk results. Interviews

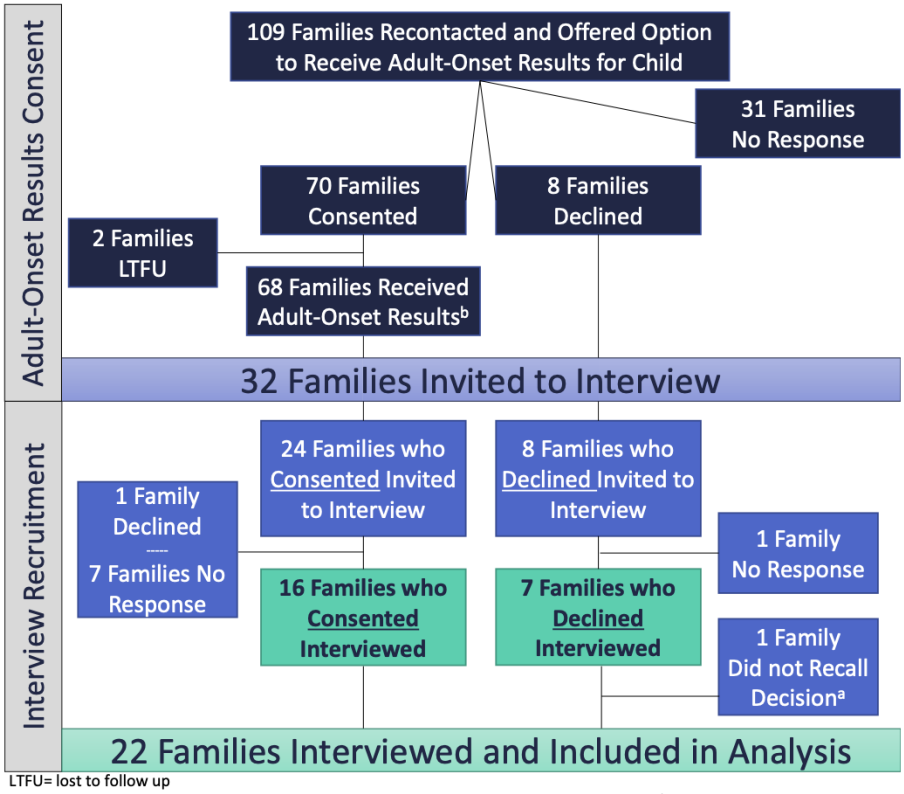
focused on how parents made the decision whether to receive these results, including their motivations to receive those results or reasons for declining them, their description of the decision-making process, and their suggestions for how to support parents in making this decision.

Interviews were audio recorded, transcribed, and stripped of identifying information. A 2-step thematic analysis process was used to identify salient themes in the data.[27] A team of 2 coders with qualitative data analysis experience (S.P. and A.M.G.) began with a broad deductive approach driven by our research questions (eg, identifying motivations for receiving and reasons for declining AO results, descriptions of the decision-making process). The coders began by coding several randomly chosen interviews together to refine the coding scheme, confirm all relevant data were being captured, and ensure both coders were interpreting and applying codes consistently. The coders then split up the remaining interviews for each to code as primary coder with the other coder as a secondary coder. Thus, all interviews were coded by both coders. Any discrepancies were identified and discussed until consensus was reached. We then used an inductive approach to abstract content from relevant segments of text to identify salient themes in a 2-step approach. The first step used an in vivo approach, privileging the interviewee's language and terminology, whereas the second step further abstracted the content into larger themes. Both coders reviewed all abstractions and used a team-based consensus approach to discuss and resolve any discrepancies in interpretation. MAXQDA qualitative analysis software (Version 2018.2.4, VERBI Software GmbH) was used to manage the coding process.

The Partners (now Mass General Brigham) Human Research Committee, the Boston Children's Hospital Institutional Review Board, and the Baylor College of Medicine Institutional Review Board approved this study. All participating parents provided verbal informed consent. The BabySeq Project was registered at ClinicalTrials.gov (NCT02422511).

Results

A total of 109 families were recontacted and offered the option to receive AO genetic results for their child in addition to their previously reported childhood-onset results (Figure 1). Of those families, 68 (62.4%) consented to receive AO results, 8 (7.3%) declined to receive adult-onset results, and 31 (28.4%) families did not respond. We invited 32 eligible families to participate in an interview and ultimately interviewed 24 parents from 23 families. Interviewed parents were 75% female with an average age of 35 years (Table 1). Although the characteristics of this cohort are generally reflective of the



LTFU= lost to follow up

[a]Parent participated in an interview but did not recall making the decision to decline results; [b]Both positive and negative

Figure 1 **Study flow diagram.**

Table 1 Parent characteristics

Characteristic	$N = 24$ (23 Families)[a]
Gender, female, *n* (%)	18 (75)
Age, mean (SD), range, y	35 (3.6), 29-44
Race and ethnicity, *n* (%)	
Asian	2 (8)
Non-Hispanic White	21 (88)
Not reported	1 (4)
Highest education level, *n* (%)	
Bachelor's degree	6 (25)
Master's degree	9 (38)
Doctoral or professional degree	5 (21)
Not reported	4 (17)
Income, *n* (%)	
$75,000 to $99,000	1 (4)
$100,000 to $149,999	5 (21)
$150,000 to $199,999	4 (17)
$200,000 to $499,999	8 (33)
Not reported	6 (25)
BabySeq Project place of enrollment, *n* (%)	
Well-baby nursery	21 (88)
ICU	3 (13)
Childhood-onset results, *n* (%)	
Positive	2 (8)
Negative	22 (92)
Adult-onset only results ($n = 17$), *n* (%)	
Positive	2 (12)
Negative	15 (88)

Not all categories sum to 100% because of missing data or rounding.
ICU, intensive care unit.
[a]Two parents participated in 1 interview together.

participants in the larger BabySeq Project, we did have more parents who identified as female participate in an interview. One parent from a family who declined to receive AO results did not recall making the decision and therefore was excluded from analysis.

Motivations for consenting to receive AO results

Parents described a number of motivations for consenting to receive results related to AO disease for their children. Each consenting family identified between 1 and 5 motivations for their decision. The most commonly cited motivation among those who consented to receive this information was the opportunity for early intervention and/or surveillance for a future disease, which was noted by 11 of 16 families who consented (Table 2). Similarly, 2 parents explained that choosing to receive AO results was part of a larger strategy of doing everything you can as a parent to protect their child (Table 2).

Implications for the family's health was another motivation for opting to receive this information, expressed by 7 families. They saw this as helpful information for themselves as parents, for their other children, for other family members, or for family planning purposes. Of note, none of these families described family benefit as the sole motivation for choosing to receive this information, but rather noted it among other reasons. One parent noted, after describing several other motivations, that this information could potentially affect her and her husband's health, "*I think it would also have been helpful if [child] had had something that maybe me or my husband would have, as well, that we could have kind of gotten a jump on it and asked about sequencing for ourselves or ask our doctors about it*" (P109, AO results negative). Another parent explained the potential benefit of learning this information for his entire family, including his other children (Table 2). Another perspective was the potential applicability of this information for family planning purposes, "*the more knowledge you have, the better it is. We want more kids, so knowing if there was anything hidden in there that we, maybe carrier genes that [child] wasn't ever showing any signs of or anything like that was going to be helpful*" (P107, AO negative).

Six families said that the ability to prepare for a future illness was a reason they consented to receive information about their child's AO disease risk. This theme was different from the parents who wanted to know this information for the purpose of early intervention and symptom surveillance. These parents instead noted that it was important for them to know simply so that they could be prepared for it, emotionally or otherwise, without mention of potential health interventions (Table 2).

Five families explained that they wanted to receive the AO information because they just generally feel that more information is better. As one mother described: "*I think we just took the idea of, the more information the better*," (P108, AO negative). Likewise, another 2 parents noted that curiosity was a motivation after describing other, more prominent motivations, with one simply stating "just curiosity" (P201, AO positive) in response to the interviewer asking if there were any other factors she considered before making the decision.

Three parents described the option to receive AO results as an extension of the original study. One parent explained that because she felt the biggest risk of participation in the BabySeq Project was the potential for discrimination, she felt that there was no additional risk to learn this other information:

> *We didn't perceive, like, a huge downside given that the sequencing data was already out there. […] For the original entry into the study I had a lot of concerns about privacy and potential for discrimination in the future about participating in the study at all. But at this point […] that was kind of already done. And so it seemed to make sense to use the data for as much good as possible.* (P106, AO negative).

Similarly, 2 families noted that contributing to research was an important reason to consent to receive this information (Table 2).

Finally, 1 parent named 2 unique motivations that were not mentioned by any other families. First, this parent saw this information as a way to support her child's autonomy:

Table 2 Parents' motivations for consenting to receive and reasons for declining adult-onset only disease risk results for their child

Theme	No. of Families Noting	Illustrative Quote
Motivations to receive AO results (n = 16 families who consented)		
Early intervention/ surveillance	11	"*We talked about it and said well, if there's anything that we can find out now that could help her in the future, make either good health choices or things that she could tell doctors to be aware of or get tested for as she grows older. We wanted to make sure she has those available.*" (P101, AO negative)
Implications for family health	7	"*I'd say yeah, the consideration, not just of her health, but also our own, mine, my wife's health, as well as the health of our parents as well. And our daughter's siblings.*" (P112, AO negative)
Ability to prepare	6	"*Here is your kid, and here is the genome, and here are the risks, actionable and non-actionable, that you're going to have as a parent. I think that lends itself to a lot of different things. It can lend itself to making financial decisions, and a whole bunch of other life decisions that you may just keep in the mind and allow you to make a more educated decision.*" (P111, AO negative)
More information is better	5	"*I'm a more knowledge is better guy.*" (P113, AO negative)
Extension of original study	3	"*It's just like a quick yes, sure, we'll be participating since she's already in the program.*" (P201, AO positive)
Contributing to research	2	"*Gosh. I think it was, again, just not having a problem with people studying us, I guess. I understand the need for sample sizes and the more you have the better. So, I figured why not.*" (P107, AO negative)
Doing everything you can	2	"*We knew that if there was any sort of thing, that we would do anything, we would go head over heels to make our child well. And that is just something that we knew that we would sell our house, do whatever. And that's the people we are. So, I think that that kind of definitely helped in the decision-making, knowing that our top priority is our children's health. And, yeah, I think that that was really the biggest factor was like, well we just want to do everything we can to keep our kids safe and healthy.*" (P103, AO negative)
Curiosity	2	"*It just seemed like a cool thing to know.*" (P109, AO negative)
Other	1	"*I think that we, at that point, felt like this is good information to have. It will give her greater autonomy to kind of make health care decisions in the future and, you know, to just give her the most choice as possible.*" (P102, AO negative)
		"*That was like around the same time I took this health care ethics class and […] as part of the class, we explored the topic and talked about it and I think, in general for me at least, it just seems to really recognize the dignity and worth of a person to give them as much information as possible.*" (P102, AO negative)
Reasons to decline AO results (n = 6 families who declined)		
Child's future autonomy	4	"*Right now, I am totally and wholly responsible for the care of my child. So, I made a choice that before he's 18 and while he's completely my responsibility, it was my choice to make about any medical decisions […]. Post 18, my initial thought was, these are* his *decisions to make, and I'm not sure if I want to impose that on him.*" (P304)
Negative impact on parenting	3	"*If we know something, and she's not able to comprehend it yet, like do we treat her differently? Do we do things differently with her?*" (P305)
Anxiety about future disease	3	"*It's tough. Do you end up living your life waiting for every little signal that something's happening? Is that it? Is it starting?*" (P306)
Negative impact on child	1	"*It might impact how she lives her life. It would definitely impact her worries and sort of anxiety about it, if indeed she had tested positive. Obviously, that's not something I want to impart on a child.*" (P303)
Genetic discrimination	1	"*We didn't want to get testing done and receive information that might affect her ability to receive health insurance in the future, premium.*" (P301; see text for full quote)
Study logistics	1	"*I think also at that time we were still feeling overwhelmed and the thought of doing another whole round of stuff [study steps] may have also affected us, our decision.*" (P306)

AO, adult-onset only results.

"*I think that we, at that point, felt like this is good information to have. It will give her greater autonomy to kind of make health care decisions in the future and, you know, to just give her the most choice as possible.*" (P102, AO negative). She also noted that part of the reason she chose to receive AO results for her child was because she felt that obtaining as much information as possible was a way of recognizing the dignity and worth of an individual: "*That was like around the same time I took this health care ethics class and […] as part of the class, we explored the topic and talked about it and I think, in general for me at least, it just seems to really recognize the dignity and worth of a person to give them as much information as possible.*" (P102, AO negative).

Reasons for declining AO results

Parents who declined to receive this information similarly noted a variety of reasons for their decision, with each family identifying between 1 and 3 reasons. The most common reason for declining identified by 4 families was the child's future autonomy. As one mother described,

> *I think that as a parent I view myself as being in charge of the health and well-being of my son because of his age and I'm able to make his medical decisions for him and I think that right kind of follows with me until the age where he can truly consent. My thought was that any adult-onset condition would occur at a time where he would be able to consent as to whether or not he wanted to know about that particular piece of information. The reason that I said no is because I felt it was kind of a violation of his right to have his own medical agency or medical decision-making*. (P30)

Three families described their concern that knowing this information could negatively impact their parenting as a reason for declining.

> *I don't want to perceive his existence any differently in terms of his ability to live, and to do stuff, and to be okay, and to fall down and get hurt, or to put a shoe in his mouth like when he did when he was [a baby], like when I got the information from the original thing [original BabySeq childhood-onset results], "He's fine." You know? I want him to be a happy, well-adjusted kid who's able to do everything that kids can do. I don't want to put him in a bubble*. (P304)

Relatedly, 3 families noted that their own anxiety about the child's potential future disease risk was a reason for declining. As one parent described, "*we don't want to be worried about it at this point in our life when she's two*" (P303). And while those parents declined because they were worried about the effect of this information on their own anxiety or the parent–child relationship, one mother also described being concerned about how knowing this information could lead to a negative effect on the child (Table 2).

One father described declining because he and his wife were concerned about the possibility of genetic discrimination, particularly within the political climate at the time:

> *The main factor was the political situation going on in health care right now, that when we made the initial decision to participate in 2016, the health law at that time was settled and preexisting conditions couldn't be penalized and we thought, oh, they'll never take away that protection. That's a wonderful thing. By 2017, with the new administration in place, threatening to just uproot everything that had happened with the health care laws, we weren't sure of that at all at that point. So, we didn't want to get testing done and receive information that might affect her ability to receive health insurance in the future, premium. […] If, say, in 2020 there was an administration that came back in and was again committed to protecting it, I'm still not sure we would make a decision to get testing because what this current administration, the Trump administration, has taught us is that things can be dismantled very quickly.* (P301)

Decision-making process

Of the 6 families who declined, 3 noted that this was a difficult decision, with some explaining that deciding whether to receive AO results was a harder decision than the original decision to participate in the BabySeq Project and receive childhood-onset results: "*Yeah, well, compared to the pediatric [results], I think it was a harder decision*," (P303, declined). Of the remaining families who declined, 2 parents did not characterize the decision as easy or difficult and the remaining family noted it was easy (Table 3). In contrast, 13 of the 16 families who consented to receive this information described the decision as easy. Of the remaining consented families, 2 described the decision as difficult and the last did not characterize it one way or the other.

Though we did not ask specifically about it, in general, parents did not describe discordant opinions when making

Table 3 Examples of parents' descriptions of ease or difficulty of decision to receive adult-onset results for their child

Consent/Decline	Easy Decision (n = 15)	Difficult Decision (n = 6)
Consenting parents	"*Super easy, 5-minute discussion. I just said 'Hey, we were asked to do this, what do you think? It's part of the BabySeq. It's more information.' And then he said 'Yeah, that sounds great, the more the better.' I think maybe it was a 2-minute conversation.*" (P108, AO negative)	"*I think we had a hard time in the very beginning, making the decision, because we were a little bit concerned about, not her specific results, but where the study was leading and if it would lead people to, I don't know. The decisions that people would make with the information, but, for us it, that was not as weighted as much as what we felt was the value of having the information.*" (P102, AO negative)
Declining parents	"*It felt easy only in, being a mom, it's so completely overwhelming. It almost felt like one more thing of like, I don't know if I want to know this right now. I'm not sure if I can think about 18 years in the future right now.*" (P304)	"*I'd say there is a benefit to knowing if you had certain issues like to worry about later on in life. So maybe that may have made it a little bit more difficult decision, but I don't know. I personally felt like I didn't want to take away her autonomy.*" (P305)

Not all families characterized the decision as easy or difficult.
AO, adult-onset only results.

the decision whether to receive these results: "*yeah, we were pretty much on the same page*," (P303, declined); "*I sort of ran it by my husband and we both agreed that we would like to know*," (P110, consented, AO negative). Other parents described following similar decision-making patterns they use for other decisions about their child's health care, relying on a primary medical decision maker: "*I think my husband defers a lot to me in these types of decisions. It was ultimately something that we discussed and came to a consensus together*," (P302, declined). However, in 2 families, parents did describe discordant opinions at some point in the decision-making process: "*I got overruled. […] we came to the decision together but that was kind of, my husband was strongly pro and I was kind of more ambivalent*," (P106, consented, AO negative). In the one interview in which both parents participated, they described not being on the same page about the decision initially but ultimately coming to agreement to decline to receive AO results to protect their child's future autonomy.

Parents' suggested support for decision-making

Parents identified several ways that researchers and/or clinicians could support parents in making the decision whether to receive AO disease risk results for their children (Table 4). The most common suggestion was to give parents more information about the conditions for which the child would be screened, including a comprehensive list of conditions, likelihood of finding risk related to one of those conditions, and what the range of anticipated clinical follow-up might be. Other related suggestions included offering such results within the context of a genetic counseling session, more time for discussion with the clinicians, and stories from parents who have received this type of result.

Discussion

In this sample of families who were offered adult-onset genetic disease risk information for their children, parents described several motivations to receive and reasons to decline this information. Those who consented to receive these results most commonly did so because they felt having this information would be beneficial to their child's and/or their family's health, whereas most parents who declined noted wanting to protect their child's future autonomy. These reasons largely mirror the debate in the literature.

Despite the longstanding push against disclosing AO genetic risk in children, our results suggest that some

Table 4 Parents' suggestions for supporting parents when deciding whether to receive adult-onset genetic disease risk results for their children ($n = 11$)

Suggested Support	Illustrative Quotes
More information on conditions screened, including list of conditions, likelihood of finding one, and what clinical follow-up would entail	"*I mean, I just, I'm very type A. I want the list [of conditions screened], and I want to know what to do with the list*," (P304, declined).
	"*I guess, maybe just making sure that the literature clearly explains what you're going to disclose, what are the actions that could result if you did know about those diseases […]*" (P303, declined).
Genetic counseling	"*I'm a big believer in genetic counselors, so just an opportunity maybe to visit with a genetic counselor I think is… I realize that that's a pretty costly thing, but that's probably like if we're talking about optimal ways to do this, make this decision, that's probably with the guidance of a real counselor*," (P113, consented, AO negative).
More discussion	"*And I guess just maybe having more than just an email conversation of do you want to go forward with this? Like having a phone call or even a meeting with parents to say, this is what we're trying to do. Maybe even this is what we'll be looking at. Would you want to know?*" (P109, consented, AO negative)
Stories from parents who have received positive AO results for their children	"*Then maybe even the experiences of other parents who have found out that their child does have these genes, and what their reactions were when they found out? And if they would go back and be like, well, if you could have changed it, do you wish you hadn't found out? Maybe that type of thing*," (P114, consented, AO negative).
Offering testing for parents if adult-onset risk is found	"*One-on-one discussion to talk through the decision if there's questions, how it could benefit the parents by knowing in the information, if it can, or even offering… say, something came back as positive. I don't know if there's room in your budget to then do some testing on the parents that might be the carrier or that might now also have it*," (P303, declined)
Help parents understand the benefit of having adult-onset information	"*I think just helping parents understand that knowing more may actually help you be able to take care of your child or other members of the family better, I think would help them kind of evaluate that decision better. Whereas I think a lot of people might knee-jerk into just fear of the unknown*" (P110, consented, AO negative)

Some of these support suggestions were already part of our protocol, including a genetic counseling session, discussion of risks and benefits and potential findings, and testing for both parents.

AO, adult-onset only results.

parents, indeed the majority in our study, do want this information, and that they consider several factors when making that decision in a way that aligns with their values. For most families, parents viewed the decision to receive AO information as part of an overall strategy to keep their child and family healthy. We have previously seen this as a reason some of the families originally chose to participate in the BabySeq Project[28] as well as a motivation of individuals to participate in other studies of genome-scale sequencing.[29,30]

Those who declined largely noted the child's future autonomy as the reason. The child's "right to an open future" has often been invoked as one of the primary reasons to not offer such results to parents.[7,31] However, as genomic sequencing has become more common, the benefits have become clearer, and empirical studies generally do not find negative psychological effect of receiving genetic information for children or their parents,[32-34] this debate has been revisited. Garrett et al[31] argue that the child's interest in an open future should not be conceived of as a right but rather an interest to weigh against other interests. Some families, like 4 of our interviewed families, may feel that their child's interest in an open future outweighs the potential benefits. However, others disagree. Notably, 1 parent who consented to receive results in our study was motivated by the idea that the results could help their child manage future health care decisions, thus supporting their child's autonomy.

Indeed, 7 families cited the benefit to their family as one of the reasons they chose to receive these results, though it was not the sole reason for any of them. Like Miner et al[20] found, some parents conceptualize AO results for their minor children as important family health information. The question of whether to consider potential benefit to the family when evaluating the best interest of the child with respect to disclosing AO genetic risk information has been a longstanding point of contention. In their 2006 statement on newborn screening, the ACMG noted that there is potential benefit for families given the genetic nature of some of the conditions tested for in standard state-run newborn screening. Indeed, in their recommendations on reporting secondary findings, the ACMG argued that the potential benefit to the parents, in addition to the future health of the child, outweighed ethical concerns about reporting predictive genetic information. While some have criticized this broader conception of the best interests of the child,[35,36] others have argued that benefit to the parent's health is a direct benefit to the child.[37]

Our participants also offered suggestions for ways to support parents in making the decision whether to receive AO results. Reassuringly for our team, several of the suggestions parents made were already part of the project protocol. Parents participated in a genetic counseling session in which the risks and potential benefits of opting to receive this information were discussed. When a positive finding was identified, parents' previously collected samples (parents provided buccal samples at enrollment for this purpose[21]) were tested for the same variant and results were disclosed to the parents. Given the time lag between consent and the interview, it is possible that some parents may have forgotten the details of the consent session. Nevertheless, it shows what was important to parents and their suggestions to provide a detailed list of conditions (vs examples) along with standard of care for each and stories from parents are novel. Although not every parent may want to receive all this additional information, being able to offer it to parents may be helpful in some cases. Genetic counselors and other clinicians may want to work on a case-by-case basis to determine what level of information individual parents desire.

Our results should be considered within the context of the study's limitations. First, the parents we interviewed had already agreed to participate in the BabySeq Project and thus were likely to hold more positive views toward genomic sequencing. Second, because this was a qualitative study, the goal of this work is not generalizability, but transferability.[38] Third, most of the parents were non-Hispanic White, with high educational attainment and high household income, limiting the transferability of these findings. Future research should explore how such decisions play out in families of different racial, ethnic, and socioeconomic backgrounds, because the values that underlie these decisions may vary. Fourth, in all but one family, we interviewed only 1 parent. We may have observed more discordant opinions between parents if we had interviewed the second parent among families with 2 parents. Finally, given the average of 18 months between making the decision and participating in the interview, parents may not have recalled the decision accurately. We did remove 1 interview from the analysis because the parent did not specifically recall making the decision. This may be because of the time lag or it may be that this was not an eventful experience for the parent within the larger BabySeq Project given he had declined AO results. He did remember specifics of the childhood-onset portion of the project.

Our findings show that parents have a wide variety of reasons for consenting or declining to receive genetic risk for AO conditions. While some families may weigh the benefits and risks and determine that receiving this information is in the best interests of their child and family, others may feel that receiving this information could do more harm than good. Our findings suggest that there are different values at stake and in tension with each other. Researchers and clinicians should provide the resources and support to promote shared decision-making and encourage families to make these choices in a way that aligns with their values.

Data Availability

Given the nature of qualitative data, the data are not available for public use. De-identified, aggregated data are available upon request.

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Ethics Declaration

The Partners (now Mass General Brigham) Human Research Committee, the Boston Children's Hospital Institutional Review Board (IRB), and the Baylor College of Medicine IRB approved this study. All participating parents provided verbal informed consent as approved by the 3 IRBs.

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Conflict of Interest

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